



## README: Genome-Wide Human SNP 5.0 Sample Data Set

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### Description of Genome-Wide Human SNP 5.0 Sample Data Set

The structure of the 44 experiments is described below. There are 10 trios comprised of 30 distinct HapMap CEPH samples. Four samples are replicated 4 times, and 1 sample is replicated three times. These samples were subjected to the Genome-Wide Human SNP 5.0 Nsp/Sty Assay according to the protocol (Affymetrix® Genome-Wide Human SNP Nsp/Sty 5.0 Assay Manual (P/N 702419)).

| Sample ID         | Gender | Family ID | Relationship         | Group | Replicates | Well Location on Sample Plate |
|-------------------|--------|-----------|----------------------|-------|------------|-------------------------------|
| NA10846           | Male   | 1334      | father               | CEPH  | 1          | A09                           |
| NA12144           | Male   | 1334      | paternal grandfather | CEPH  | 1          | A08                           |
| NA12145           | Female | 1334      | paternal grandmother | CEPH  | 1          | A05                           |
| NA07019           | Female | 1340      | mother               | CEPH  | 1          | B05                           |
| NA07022           | Male   | 1340      | maternal grandfather | CEPH  | 1          | A04                           |
| NA07056           | Female | 1340      | maternal grandmother | CEPH  | 1          | A02                           |
| NA06985           | Female | 1341      | maternal grandmother | CEPH  | 1          | D10                           |
| NA06991           | Female | 1341      | mother               | CEPH  | 1          | D01                           |
| NA06993           | Male   | 1341      | maternal grandfather | CEPH  | 4          | B01, B11, C11, D04            |
| NA07345           | Female | 1345      | maternal grandmother | CEPH  | 1          | C04                           |
| NA07348           | Female | 1345      | mother               | CEPH  | 1          | D11                           |
| NA07357           | Male   | 1345      | maternal grandfather | CEPH  | 4          | A07, B03, C01, D09            |
| NA10855           | Female | 1350      | mother               | CEPH  | 1          | D06                           |
| NA11831           | Male   | 1350      | maternal grandfather | CEPH  | 1          | C08                           |
| NA11832           | Female | 1350      | maternal grandmother | CEPH  | 4          | A10, B08, C02, D07            |
| NA10831           | Female | 1408      | mother               | CEPH  | 1          | B06                           |
| NA12155           | Male   | 1408      | maternal grandfather | CEPH  | 1          | A11                           |
| NA12156           | Female | 1408      | maternal grandmother | CEPH  | 1          | B02                           |
| NA10835           | Male   | 1416      | father               | CEPH  | 1          | D05                           |
| NA12248           | Male   | 1416      | paternal grandfather | CEPH  | 1          | D02                           |
| NA12249           | Female | 1416      | paternal grandmother | CEPH  | 3          | B07, B10, C05                 |
| NA12740           | Female | 1444      | mother               | CEPH  | 1          | C06                           |
| NA12750           | Male   | 1444      | maternal grandfather | CEPH  | 1          | D08                           |
| NA12751           | Female | 1444      | maternal grandmother | CEPH  | 1          | C07                           |
| NA12753           | Female | 1447      | mother               | CEPH  | 1          | A01                           |
| NA12762           | Male   | 1447      | maternal grandfather | CEPH  | 1          | B04                           |
| NA12763           | Female | 1447      | maternal grandmother | CEPH  | 1          | A06                           |
| NA12801           | Male   | 1454      | father               | CEPH  | 4          | A03, B09, C03, C09            |
| NA12812           | Male   | 1454      | paternal grandfather | CEPH  | 1          | C10                           |
| NA12813           | Female | 1454      | paternal grandmother | CEPH  | 1          | D03                           |
| Total Experiments |        |           |                      |       |            | 44                            |

Table 1 – Description of 44 Samples

| CEL Filename                        | Sample  | SampleID | FamilyID | Gender | FatherID | MotherID |
|-------------------------------------|---------|----------|----------|--------|----------|----------|
| NA10846_Op1_011206_VnV_A09_r1.CEL   | NA10846 | 1        | 1334     | Male   | 2        | 3        |
| NA12144_Op1_011206_VnV_A08_r1.CEL   | NA12144 | 2        | 1334     | Male   | 0        | 0        |
| NA12145_Op1_011206_VnV_A05_r1.CEL   | NA12145 | 3        | 1334     | Female | 0        | 0        |
| NA07019_Op1_011206_VnV_B05_r1.CEL   | NA07019 | 4        | 1340     | Female | 5        | 6        |
| NA07022_Op1_011206_VnV_A04_r1.CEL   | NA07022 | 5        | 1340     | Male   | 0        | 0        |
| NA07056_Op1_011206_VnV_A02_r1.CEL   | NA07056 | 6        | 1340     | Female | 0        | 0        |
| NA06985_Op1_011206_VnV_D10_r1.CEL   | NA06985 | 7        | 1341     | Female | 0        | 0        |
| NA06991_Op1_011206_VnV_D01_r1.CEL   | NA06991 | 8        | 1341     | Female | 9        | 7        |
| NA06993_Op1_011206_VnV_B01_r1.CEL   | NA06993 | 9        | 1341     | Male   | 0        | 0        |
| NA06993_Op1_011206_VnV_B11_r1.CEL   | NA06993 | 9        | 1341     | Male   | 0        | 0        |
| NA06993_Op1_011206_VnV_C11_r1.CEL   | NA06993 | 9        | 1341     | Male   | 0        | 0        |
| NA06993_Op1_011206_VnV_D04_r1.CEL   | NA06993 | 9        | 1341     | Male   | 0        | 0        |
| NA07345_Op1_011206_VnV_C04_r1.CEL   | NA07345 | 10       | 1345     | Female | 0        | 0        |
| NA07348_Op1_011206_VnV_D11_r1.CEL   | NA07348 | 11       | 1345     | Female | 12       | 10       |
| NA07357_Op1_011206_VnV_A07_r1.CEL   | NA07357 | 12       | 1345     | Male   | 0        | 0        |
| NA07357_Op1_011206_VnV_B03_r1.CEL   | NA07357 | 12       | 1345     | Male   | 0        | 0        |
| NA07357_Op1_011206_VnV_C01_r1.CEL   | NA07357 | 12       | 1345     | Male   | 0        | 0        |
| NA07357_Op1_011206_VnV_D09_r1.CEL   | NA07357 | 12       | 1345     | Male   | 0        | 0        |
| NA10855_Op1_011206_VnV_D06_r1.CEL   | NA10855 | 13       | 1350     | Female | 14       | 15       |
| NA11831_Op1_011206_VnV_C08_r1.CEL   | NA11831 | 14       | 1350     | Male   | 0        | 0        |
| NA11832_Op1_011206_VnV_A10_r1.CEL   | NA11832 | 15       | 1350     | Female | 0        | 0        |
| NA11832_Op1_011206_VnV_B08_r1.CEL   | NA11832 | 15       | 1350     | Female | 0        | 0        |
| NA11832_Op1_011206_VnV_C02_r1.CEL   | NA11832 | 15       | 1350     | Female | 0        | 0        |
| NA11832_Op1_011206_VnV_D07_r1.CEL   | NA11832 | 15       | 1350     | Female | 0        | 0        |
| NA10831_Op1_011206_VnV_B06_r1.CEL   | NA10831 | 16       | 1408     | Female | 17       | 18       |
| NA12155_Op1_011206_VnV_A11_r1.CEL   | NA12155 | 17       | 1408     | Male   | 0        | 0        |
| NA12156_Op1_011206_VnV_B02_r1.CEL   | NA12156 | 18       | 1408     | Female | 0        | 0        |
| NA10835_Op1_011206_VnV_D05_r1.CEL   | NA10835 | 19       | 1416     | Male   | 20       | 21       |
| NA12248_Op1_011206_VnV_D02_r1.CEL   | NA12248 | 20       | 1416     | Male   | 0        | 0        |
| NA12249_Op1_011206_VnV_B07_r1.CEL   | NA12249 | 21       | 1416     | Female | 0        | 0        |
| NA12249_Op1_011206_VnV_B10_r1.CEL   | NA12249 | 21       | 1416     | Female | 0        | 0        |
| NA12249_Op1_011206_VnV_C05_r1.CEL   | NA12249 | 21       | 1416     | Female | 0        | 0        |
| NA12740_Op1_011206_VnV_C06_r1.CEL   | NA12740 | 22       | 1444     | Female | 23       | 24       |
| NA12750_Op1_011206_VnV_D08_r1.CEL   | NA12750 | 23       | 1444     | Male   | 0        | 0        |
| NA12751_Op1_011206_VnV_C07_r1.CEL   | NA12751 | 24       | 1444     | Female | 0        | 0        |
| NA12753_Op1_011206_VnV_A01_r1.CEL   | NA12753 | 25       | 1447     | Female | 26       | 27       |
| NA12762_Op1_011206_VnV_B04_r1.CEL   | NA12762 | 26       | 1447     | Male   | 0        | 0        |
| NA12763_Op1_011206_VnV_A06_r1.CEL   | NA12763 | 27       | 1447     | Female | 0        | 0        |
| NA12801_Op1_011206_VnV_A03_r1_2.CEL | NA12801 | 28       | 1454     | Male   | 29       | 30       |
| NA12801_Op1_011206_VnV_B09_r1.CEL   | NA12801 | 28       | 1454     | Male   | 29       | 30       |
| NA12801_Op1_011206_VnV_C03_r1.CEL   | NA12801 | 28       | 1454     | Male   | 29       | 30       |
| NA12801_Op1_011206_VnV_C09_r1.CEL   | NA12801 | 28       | 1454     | Male   | 29       | 30       |
| NA12812_Op1_011206_VnV_C10_r1.CEL   | NA12812 | 29       | 1454     | Male   | 0        | 0        |
| NA12813_Op1_011206_VnV_D03_r1.CEL   | NA12813 | 30       | 1454     | Female | 0        | 0        |

**Table 2 – Pedigree Information**

## Results

Using the BRLMM-P algorithm at the default setting of 0.05, the average call rate for these samples was 99.65% and the concordance with HapMap genotypes was 99.70%. The Mendelian inheritance consistency was found to be 99.96%. Reproducibility was measured at 99.9%.

Figure 1 show the concordance relative to HapMap release 20. The values are averaged over the 44 experiments, and the dashed lines are the bootstrap 95% confidence intervals. The CRC curve is based on the default 440K SNPs.

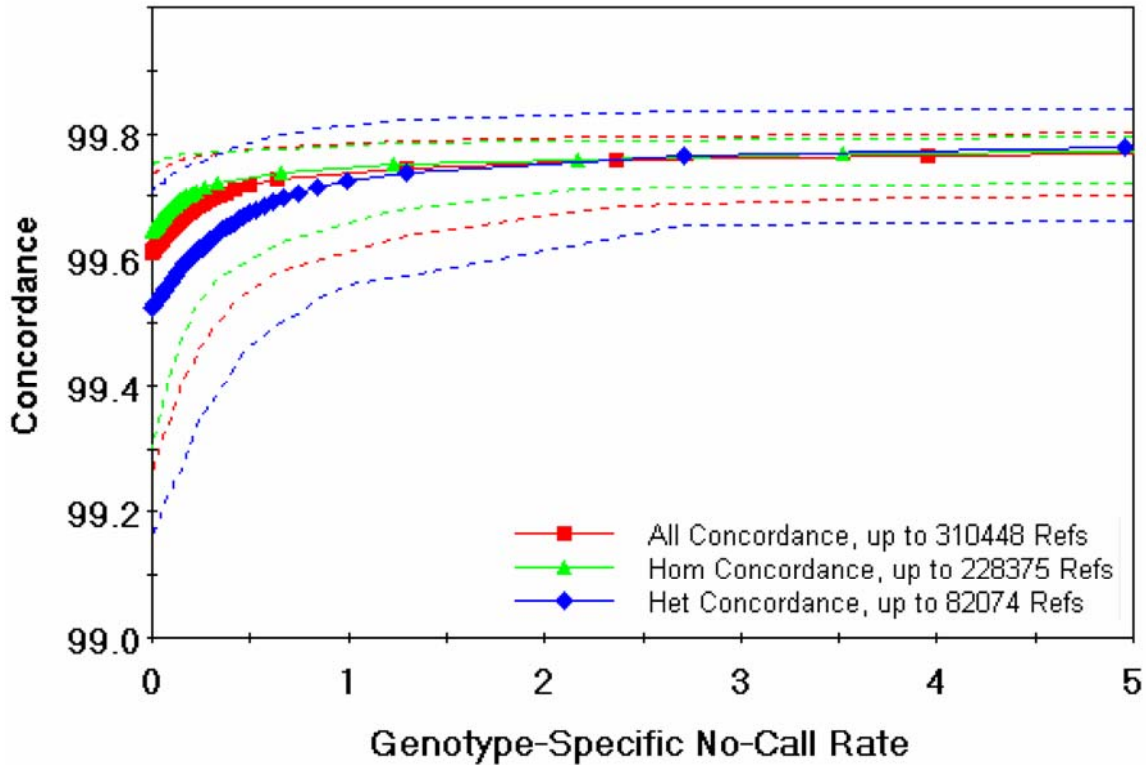


Figure 1 – No call rate versus concordance plot

### Download Information

The data consists of GCOS CEL and XML files as well as Command Console CEL, CHP, and ARR files. The available options for download are:

- 1) GCOS CEL files.
  - GW\_SNP5\_GCOS\_CEL\_1.zip (417 MB)
  - GW\_SNP5\_GCOS\_CEL\_2.zip (412 MB)
- 2) GCOS XML files.
  - GW\_SNP5\_GCOS\_XML.zip (132 KB)
    - The XML files contain the pedigree information for these samples as displayed in Table 2.
- 3) AGCC CEL files.
  - GW\_SNP5\_AGCC\_CEL\_1.zip (382 MB)
  - GW\_SNP5\_AGCC\_CEL\_2.zip (378 MB)
    - Affymetrix GeneChip Command Console™ Software (AGCC) will be the replacement for the GeneChip Operating Software (GCOS).  
[http://www.affymetrix.com/products/software/specific/command\\_console\\_software.affx](http://www.affymetrix.com/products/software/specific/command_console_software.affx)
- 4) AGCC CHP files.
  - GW\_SNP5\_AGCC\_CHP\_1.zip (125 MB)
  - GW\_SNP5\_AGCC\_CHP\_2.zip (125 MB)

- Information about the AGCC CHP file format can be found here:  
<http://www.affymetrix.com/support/developer/index.affx>
- 5) AGCC ARR files.
- GW\_SNP5\_AGCC\_ARR.zip (41 KB)
    - The AGCC software will store the sample attributes such as pedigree information (Table 2) in the ARR files.

### **Additional Information and Support**

Please refer to the complete Affymetrix® Genome-Wide Human SNP Nsp/Sty 5.0 Assay Manual for additional information. For support information, please contact your FAS or local support team

<http://www.affymetrix.com/support/index.affx>

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